



## harlequin ichthyosis

Harlequin ichthyosis is a severe genetic disorder that mainly affects the skin. Infants with this condition are born with very hard, thick skin covering most of their bodies. The skin forms large, diamond-shaped plates that are separated by deep cracks (fissures). These skin abnormalities affect the shape of the eyelids, nose, mouth, and ears, and limit movement of the arms and legs. Restricted movement of the chest can lead to breathing difficulties and respiratory failure.

The skin normally forms a protective barrier between the body and its surrounding environment. The skin abnormalities associated with harlequin ichthyosis disrupt this barrier, making it more difficult for affected infants to control water loss, regulate their body temperature, and fight infections. Infants with harlequin ichthyosis often experience an excessive loss of fluids (dehydration) and develop life-threatening infections in the first few weeks of life. It used to be very rare for affected infants to survive the newborn period. However, with intensive medical support and improved treatment, people with this disorder now have a better chance of living into childhood and adolescence.

### Frequency

Harlequin ichthyosis is very rare; its exact incidence is unknown.

### Genetic Changes

Mutations in the *ABCA12* gene cause harlequin ichthyosis. The *ABCA12* gene provides instructions for making a protein that is essential for the normal development of skin cells. This protein plays a major role in the transport of fats (lipids) in the outermost layer of skin (the epidermis). Some mutations in the *ABCA12* gene prevent the cell from making any ABCA12 protein. Other mutations lead to the production of an abnormally small version of the protein that cannot transport lipids properly. A loss of functional ABCA12 protein disrupts the normal development of the epidermis, resulting in the hard, thick scales characteristic of harlequin ichthyosis.

### Inheritance Pattern

This condition is inherited in an autosomal recessive pattern, which means both copies of the gene in each cell have mutations. The parents of an individual with an autosomal recessive condition each carry one copy of the mutated gene, but they typically do not show signs and symptoms of the condition.

## **Other Names for This Condition**

- Harlequin baby syndrome
- HI
- Ichthyosis Congenita, Harlequin Fetus Type

## **Diagnosis & Management**

### Genetic Testing

- Genetic Testing Registry: Autosomal recessive congenital ichthyosis 4B  
<https://www.ncbi.nlm.nih.gov/gtr/conditions/C0239849/>

### Other Diagnosis and Management Resources

- GeneReview: Autosomal Recessive Congenital Ichthyosis  
<https://www.ncbi.nlm.nih.gov/books/NBK1420>

### General Information from MedlinePlus

- Diagnostic Tests  
<https://medlineplus.gov/diagnostictests.html>
- Drug Therapy  
<https://medlineplus.gov/drugtherapy.html>
- Genetic Counseling  
<https://medlineplus.gov/geneticcounseling.html>
- Palliative Care  
<https://medlineplus.gov/palliativecare.html>
- Surgery and Rehabilitation  
<https://medlineplus.gov/surgeryandrehabilitation.html>

## **Additional Information & Resources**

### MedlinePlus

- Health Topic: Skin Conditions  
<https://medlineplus.gov/skinconditions.html>

### Genetic and Rare Diseases Information Center

- Harlequin ichthyosis  
<https://rarediseases.info.nih.gov/diseases/6568/harlequin-ichthyosis>

### Educational Resources

- Disease InfoSearch: Harlequin Ichthyosis  
<http://www.diseaseinfosearch.org/Harlequin+Ichthyosis/3242>
- National Registry for Ichthyosis and Related Disorders, University of Washington  
<http://depts.washington.edu/ichreg/ichthyosis.registry/>
- Orphanet: Harlequin ichthyosis  
[http://www.orpha.net/consor/cgi-bin/OC\\_Exp.php?Lng=EN&Expert=457](http://www.orpha.net/consor/cgi-bin/OC_Exp.php?Lng=EN&Expert=457)

### Patient Support and Advocacy Resources

- Foundation for Ichthyosis and Related Skin Types (FIRST)  
[http://www.firstskinfoundation.org/content.cfm/category\\_id/741/page\\_id/547](http://www.firstskinfoundation.org/content.cfm/category_id/741/page_id/547)
- National Organization for Rare Disorders (NORD)  
<https://rarediseases.org/rare-diseases/ichthyosis-harlequin-type/>
- Resource list from the University of Kansas Medical Center  
<http://www.kumc.edu/gec/support/ichthyos.html>

### GeneReviews

- Autosomal Recessive Congenital Ichthyosis  
<https://www.ncbi.nlm.nih.gov/books/NBK1420>

### ClinicalTrials.gov

- ClinicalTrials.gov  
<https://clinicaltrials.gov/ct2/results?cond=%22harlequin+ichthyosis%22>

### Scientific Articles on PubMed

- PubMed  
<https://www.ncbi.nlm.nih.gov/pubmed?term=%28harlequin+ichthyosis%5BTIAB%5D%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+2520+days%22%5Bdp%5D>

### OMIM

- ICHTHYOSIS, CONGENITAL, AUTOSOMAL RECESSIVE 4B  
<http://omim.org/entry/242500>

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